

NC RARE DISEASE ADVISORY COUNCIL

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North Carolina Rare Disease Advisory Council Meeting Thursday, January 9, 2020 IQVIA Biotech 1700 Perimeter Park Drive Morrisville, NC 27560

1:00 -1:10	Welcome, Introductions of Guests	Bruce Cairns
1:30 – 1:40	Gene Therapy Updates Genetics Updates	Aravind Asokan Cindy Powell
1:40 – 1:50	Patient Advocacy Story Piedmont Health Services	Justina Williams
1:50 – 2:00	Task Force Reports	Stephen & Sandi Lehrman
2:00 – 2:15	NC TraCS Institute	Tim Carey, Mike Knowles Bruce Cairns
2:15 – 2:25	Undiagnosed Network Updates	Vandana Shashi
2:30 – 2:35	Legislative Updates & Rare Disease Day	Rufus Edmisten Tara Britt
2:35 – 3:00	2020 Focus & Future Meetings	Council

Attendees:

Council Members: Bruce Cairns, Vandana Shashi, Michael Knowles, Rufus Edmisten, Brenda Nielsen, Aravind Asokan, Stephen Lehrman, Tara Britt.

Guests:

Tim Carey: NC TraCS

Lee Ann Lawson, Michael Losow: Amicus

Jerry Walter: Fabry

Nick Kenny, Mallik Mathuri: Syneos Health

Allison Grannis: Pfizer

Kirby Consier: Novartis

Susan Nichols: Falcon Therapeutics and Rare Disease Innovations Institute

Ali Smyth, Raymond Cook, Sarah Crommey: IQVIA

Sandi Lehrman: Council Task force

Becky Sansbury: Rare Disease Innovations Institute

Cindy Powell: UNC

Mary Ann Naegle: Centogene

Erin Frey: Cure Duchenne

Terri Klein: MPS

Justina Williams: Piedmont Sickle Cell Agency

Minutes:

Bruce and Tara welcomed everyone and Tara proceeded with group introductions.

Aravind Asokan presented the gene therapy updates. Critical mass in GT growth higher than the last decade. Aravind discussed technology and science as well as clinical trials in gene therapy.

GMP manufacturing is the bottleneck and continues to be an issue.

Critical to establish links between industry, academics and advocacy.

Question on how Council could help to address the urgency for gene therapy.

Aravind said it took 20 years for GT to be commercialized. Gene Editing will be faster.

Council can be an information engine regarding this, connecting the dots between newborn screening and the undiagnosed disease network to pre-clinical discovery. Bridge gaps from discovery to therapeutics with FDA interaction.

Separate ultra-rare from rare.

Cindy Powell gave the genetics update. Cindy reported that the Newborn Screening Saves Lives Reauthorization Act of 2019 has not passed the U.S. Senate. Department of Health and Human Services Secretary Azur received a letter of support for the legislation from several Rare Disease patient advocacy groups. Cindy hopes that the Secretary will make the ACHDNC a temporary committee until the reauthorization bill is passed.

Cindy discussed the Ending Diagnostic Odyssey Act that would allow States to pay for whole genome sequencing for children on Medicaid.

Justina Williams was our guest speaker representing the Sickle Cell community. Justina shared her own personal story living with sickle cell and working with Piedmont Health

Services and Sickle Cell Agency. She discussed the importance of advocacy as well as medical education and education therapies.

Tim Carey discussed the NC TraCS program and how UNC, Duke and Wake Forest are a part of NIH Clinical and Translational Science Awards (CTSA) Network. He mentioned these CTSA's would like to join our Council meetings to see more how we could work together. Mike Knowles and Bruce also discussed the importance of this program.

Vandana Shashi talked about the UDN at Duke. She also talked about repurposing drugs to treat rare diseases and finding the appropriate resources for patients.

Tara and Rufus reviewed the NC State legislature problems with the budget which prevented the Council from receiving the 250k requested to expand the Council networks. Group discussion is that the Council would do this without current funding.

Tara talked about Rare Disease Day events. IQVIA is having a rare disease day event on February 27th.

Syneos Health in collaboration with Rare Disease Innovations Institute are having the 2nd annual rare disease day event on February 27th from 11-3.

NIH Rare Disease Day on February 28th with several members of our community participating.

Stephen Lehrman talked about task force reports, white papers and what issues the Council wants to focus on for 2020. The Council asked the Task Force to develop two reports on The Needs of Rare Disease Patients in Rural North Carolina and Cell and Gene Therapy Manufacturing Capability in North Carolina with the possibility North Carolina could take a national leadership position on these advance manufacturing technologies.

There was a lot of open discussion with council members and visitors regarding rare disease.

Bruce brought up a motion to add another council member as a subject matter expert to continue to lead the task force reports but also to focus on expanding our council to the rural parts of the state. The motion was for Stephen Lehrman to be added as a council member and the vote was unanimous to add him to our council to expand on work currently being addressed with task force and to also help lead the rural council expansion efforts.